MAYO CLINIC LABORATORIES

Test Definition: GCT

Galactosemia Reflex, Blood

Reporting Title: Galactosemia Reflex, B
Performing Location: Rochester

Ordering Guidance:

This test is appropriate for the diagnosis of, and routine carrier screening for, galactose-1-phosphate uridyltransferase deficiency.

This assay is **not appropriate** for monitoring dietary compliance. For dietary monitoring, order GAL1P / Galactose-1-Phosphate, Erythrocytes.

Necessary Information:

Patient's age is required.

Specimen Requirements:

Multiple whole blood tests for galactosemia can be performed on one specimen. Prioritize order of testing when submitting specimens. For a list of tests that can be ordered together, see Galactosemia-Related Test List.

Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: Green top (sodium heparin) or yellow top (ACD)
Specimen Volume: 5 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot**.

Forms:

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. If not ordering electronically, complete, print, and send an Biochemical Genetics Test Request (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole Blood EDTA	Refrigerated (preferred)	28 days	
	Ambient	14 days	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
8333	Gal-1-P Uridyltransferase, RBC	Numeric	nmol/h/mg Hb	24082-0
2296	Interpretation (GALT)	Alphanumeric		59462-2
58115	Reviewed By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82775

81406 (if appropriate)

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
GALZ	Galactosemia, Full Gene Analysis	1	81406	No	Yes
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
GALZ	608596	Test Description	Alphanumeric		62364-5
GALZ	608597	Specimen	Alphanumeric		31208-2
GALZ	608598	Source	Alphanumeric		31208-2
GALZ	608599	Result Summary	Alphanumeric		50397-9
GALZ	608600	Result	Alphanumeric		82939-0
GALZ	608601	Interpretation	Alphanumeric		69047-9
GALZ	608602	Resources	Alphanumeric		99622-3
GALZ	608603	Additional Information	Alphanumeric		48767-8
GALZ	608604	Method	Alphanumeric		85069-3
GALZ	608605	Genes Analyzed	Alphanumeric		48018-6
GALZ	608606	Disclaimer	Alphanumeric		62364-5
GALZ	608607	Released By	Alphanumeric		18771-6

Reference Values:

> or =24.5 nmol/h/mg of hemoglobin